



POSNA

The Core Curriculum

Sex linked dominant disorders

Objectives

1. Describe the features of sex-linked inheritance
2. Describe the method of inheritance of sex-linked dominant disorders, and the characteristics of a pedigree of a sex-linked dominant disorder
3. List 2 sex-linked dominant disorders with orthopaedic importance

Discussion

Sex-linked disorders are a result of gene mutations located on the X chromosome. A characteristic of X linked dominant inheritance is that all of the daughters and none of the sons of an affected male are affected. Affected females transmit the disorder to one half of their children, regardless of sex (since the mother contributes one X chromosome to all children, the father only to daughters). There are only 2 conditions with this type of inheritance seen by the orthopaedic surgeon, hypophosphatemic (or Vitamin D resistant) rickets and Rett's syndrome.1. Rett syndrome is lethal in males at birth.

References

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2. Horan F, Beighton P. Orthopaedic problems in inherited musculoskeletal disorders. Berlin: Springer-Verlag; 1982.
3. Jorde LB, Carey JC, White RL. Medical Genetics. 2 ed. St. Louis: Mosby; 1999.